

# Adolescent idiopathic scoliosis

## **Description**

Adolescent idiopathic scoliosis is an abnormal curvature of the spine that appears in late childhood or adolescence. Instead of growing straight, the spine develops a side-to-side curvature, usually in an elongated "S" or "C" shape; the bones of the spine are also slightly twisted or rotated.

Adolescent idiopathic scoliosis appears during the adolescent growth spurt, a time when children are growing rapidly. In many cases the abnormal spinal curve is stable, although in some children the curve is progressive (meaning it becomes more severe over time). For unknown reasons, severe and progressive curves occur more frequently in girls than in boys. However, mild spinal curvature is equally common in girls and boys.

Mild scoliosis generally does not cause pain, problems with movement, or difficulty breathing. It may only be diagnosed if it is noticed during a regular physical examination or a scoliosis screening at school. The most common signs of the condition include a tilt or unevenness (asymmetry) in the shoulders, hips, or waist, or having one leg that appears longer than the other. A small percentage of affected children develop more severe, pronounced spinal curvature.

Scoliosis can occur as a feature of other conditions, including a variety of genetic syndromes. However, adolescent idiopathic scoliosis typically occurs by itself, without signs and symptoms affecting other parts of the body.

# Frequency

Adolescent idiopathic scoliosis is the most common spinal abnormality in children. It affects an estimated 2 to 3 percent of children in the U.S.

### **Causes**

The term "idiopathic" means that the cause of this condition is unknown. Adolescent idiopathic scoliosis probably results from a combination of genetic and environmental factors. Studies suggest that the abnormal spinal curvature may be related to hormonal problems, abnormal bone or muscle growth, nervous system abnormalities, or other factors that have not been identified.

Researchers suspect that many genes are involved in adolescent idiopathic scoliosis. Some of these genes likely contribute to causing the disorder, while others play a role in determining the severity of spinal curvature and whether the curve is stable or progressive. Although many genes have been studied, few clear and consistent genetic associations with adolescent idiopathic scoliosis have been identified.

#### Inheritance

Adolescent idiopathic scoliosis can be sporadic, which means it occurs in people without a family history of the condition, or it can cluster in families. The inheritance pattern of adolescent idiopathic scoliosis is unclear because many genetic and environmental factors appear to be involved. However, having a close relative (such as a parent or sibling) with adolescent idiopathic scoliosis increases a child's risk of developing the condition.

#### Other Names for This Condition

- AIS
- Late onset idiopathic scoliosis

#### **Additional Information & Resources**

### **Genetic Testing Information**

Genetic Testing Registry: Scoliosis, idiopathic 3 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1837461/)

# Genetic and Rare Diseases Information Center

 Adolescent idiopathic scoliosis (https://rarediseases.info.nih.gov/diseases/552/adole scent-idiopathic-scoliosis)

#### Patient Support and Advocacy Resources

- Disease InfoSearch (https://www.diseaseinfosearch.org/)
- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

### Research Studies from ClinicalTrials.gov

 ClinicalTrials.gov (https://clinicaltrials.gov/search?cond="Adolescent idiopathic scoliosis")

### Catalog of Genes and Diseases from OMIM

- SCOLIOSIS, ISOLATED, SUSCEPTIBILITY TO, 1 (https://omim.org/entry/181800)
- SCOLIOSIS, ISOLATED, SUSCEPTIBILITY TO, 2 (https://omim.org/entry/607354)
- SCOLIOSIS, ISOLATED, SUSCEPTIBILITY TO, 3 (https://omim.org/entry/608765)
- SCOLIOSIS, ISOLATED, SUSCEPTIBILITY TO, 4 (https://omim.org/entry/612238)
- SCOLIOSIS, ISOLATED, SUSCEPTIBILITY TO, 5 (https://omim.org/entry/612239)

#### Scientific Articles on PubMed

 PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Scoliosis%5BMAJR%5D%29 +AND+%28adolescent+idiopathic+scoliosis%5BTIAB%5D%29+AND+%28gene+OR +genetic%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1 800+days%22%5Bdp%5D)

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